

Teaching NeuroImages: Coats disease revealing facioscapulohumeral muscular dystrophy

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A 21-year-old Brazilian woman presented with mild facial muscle weakness, scapular winging and an asymmetric shoulder girdle, and peroneal weakness. Medical history disclosed mildly symptomatic Coats disease (CD) (figure). Muscle MRI studies were highly suggestive of facioscapulohumeral muscular dystrophy type 1 (FSHD1) (figure) and genetic testing confirmed this diagnosis.

FSHD1 represents an autosomal dominant muscular dystrophy with typical muscle MRI patterns associated with extramuscular manifestations, including sensorineural deafness and complex retinal vasculopathy with telangiectasis, such as CD.¹ CD may be a presenting feature of FSHD1 with high risk of exudative retinal detachment, retinal microaneurysm formation, and peripheral retinal telangiectasis.²

AUTHOR CONTRIBUTIONS

P.V.S. Souza: case report project: conception, organization, execution; manuscript: writing of the first draft. W.B.V.R. Pinto: case

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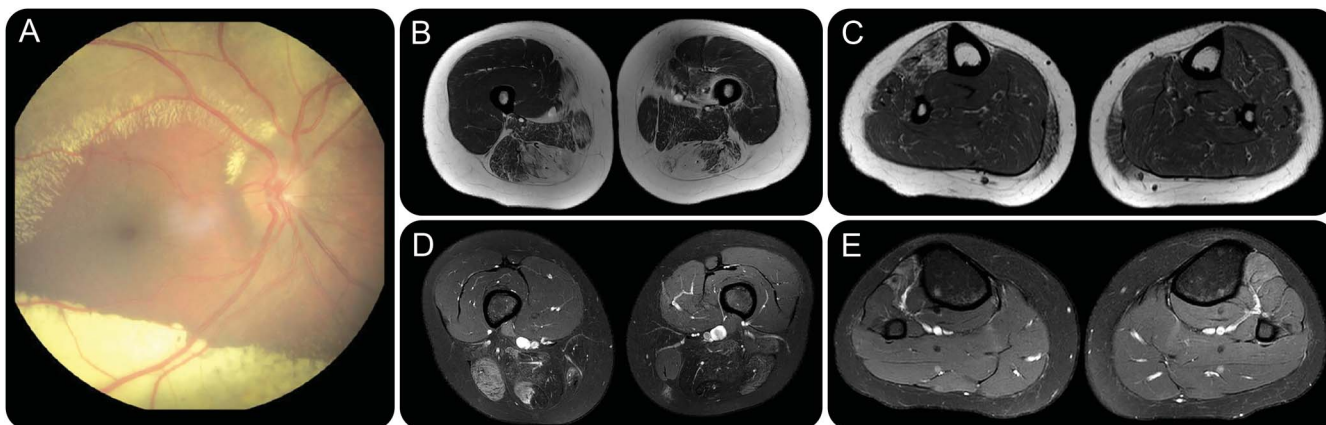
DISCLOSURE

The authors report no disclosures relevant to the manuscript. Go to Neurology.org for full disclosures.

REFERENCES

1. Leung DG, Carrino JA, Wagner KR, Jacobs MA. Whole-body magnetic resonance imaging evaluation of facioscapulohumeral muscular dystrophy. *Muscle Nerve* 2015;52:512–520.
2. Statland JM, Sacconi S, Farmakidis C, Donlin-Smith CM, Chung M, Tawil R. Coats syndrome in facioscapulohumeral dystrophy type 1: frequency and D4Z4 contraction size. *Neurology* 2013;80:1247–1250.

Figure Fundusoscopic evaluation and muscle MRI studies in facioscapulohumeral muscular dystrophy



(A) Fundusoscopic analysis showing retinal atrophy and massive exudate deposition in peripheral retina. Axial muscle MRI disclosing fatty replacement in posterior thigh muscles (semimembranosus and semitendinosus) and in right anterior tibial compartment in T1-weighted sequences (B, C) and T2-weighted sequences with fat suppression (D, E).

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